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ABSTRACT

This information sheet briefly describes the history of the identification of Down Syndrome, its prenatal diagnosis, characteristics of individuals with Down Syndrome, its causes, its rate of occurrence and recurrence, its impact on child development, and recommended content of education programs for children with Down Syndrome. A list of seven organizational and bibliographic resources is included. (JDD)

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DOWN SYNDROME

Background

The term Down syndrome is taken from the name of the English physician, Dr. John Langdon Down, who is credited with first describing the condition in 1866. It was not until 1959 that the actual chromosomal abnormality associated with the syndrome was discovered. Dr. Jerome Lejuene found that individuals with Down syndrome possessed additional genetic material in their cells, usually an extra chromosome. Instead of having 46 chromosomes in each cell (22 pair of autosomes or non-sex chromosomes and one pair of sex chromosomes, xx in females, xy in males), individuals with Down syndrome most commonly have 47 chromosomes with the extra chromosome associated with the 21st pair. The term Trisomy 21 is therefore used to describe this configuration of three #21 chromosomes. About 95% of all individuals with Down syndrome have Trisomy 21.

What Are the Characteristics of Individuals with Down Syndrome?

Individuals with Down syndrome may vary significantly in terms of physical and psychological characteristics. The list of possible characteristics however should not obscure two important facts: clearly individuals with Down syndrome are first and foremost people who have similar needs, desires, and rights as others; and, the effects of intensive interventions with young Down syndrome children are only now being evaluated, hence making many of the historical descriptions of Down syndrome no longer accurate.

Some of the physical characteristics observed in persons with Down syndrome include the following: the back of the head is often flattened, the eyelids may be slightly slanted, small skin folds at the inner corners of the eyes may be present, the nasal bridge is slightly depressed, and the nose and ears are usually somewhat smaller. In the newborn there is often an excess of skin at the back of the neck. The hands and feet are small and the fingerprints are often different from chromosomally normal children.

Individuals with Down syndrome have loose ligaments and their muscle strength and tone are usually reduced. If the ligaments between the first two neck bones are loose, there may be a condition referred to as Atlanto-Axial Instability. About one-third of children with Down syndrome have congenital heart disease. Other congenital defects such as a blockage in the bowels and cataracts, although rare, may also be present. Hearing deficits, visual problems, and thyroid dysfunction are often observed in persons with Down syndrome.

What Causes Down Syndrome?

In Trisomy 21, the extra chromosome can usually be traced to the egg, although it has been hypothesized that it may come from the sperm in 20% of the cases. It is suspected that during cell division of the egg (or sperm), the two #21 chromosomes do not separate properly and one cell will therefore have an extra #21 chromosome which later, if united with a normal cell, may lead to Trisomy 21. What causes this improper cell division is unknown. Factors such as viral infections, hormonal abnormalities, x-rays, and certain drugs are being investigated as potential causal factors. A small number of cases are genetic in origin; these can be determined through genetic counseling.

How Often Does Down Syndrome Occur/Recur?

Down syndrome is the most common clinical cause of mental retardation in the world. It occurs once in every 1,500–2,000 births when the mother's age is below 25; one in every 400 births when the mother's age is over 35; once in every 40 births when the mother's age is over 45. A mother with a child with Down syndrome has a one in 25 chance of recurrence. Siblings of Down syndrome have the same risk of having a child with Down syndrome as does the general population unless the disorder is of genetic origin.

How Does Down Syndrome Affect Development?

All areas of development may be delayed in a child with Down syndrome. Poor muscle tone influences gross and fine motor development but this can be improved with physical therapy; a consistent, structured program of physical activity; and an ongoing weight maintenance plan. Likewise, language development delays due to muscle problems and cognitive limitations can be alleviated via structured stimulation programs and language therapy to improve skills.

Since mental retardation frequently occurs in children with Down syndrome, higher integrative abilities such as the ability to think abstractly and to form concepts are likely to be affected. However, appropriate educational programs have demonstrated impressive successes in teaching functional academic skills as well as critical self-help and daily living skills. Most individuals with Down syndrome learn to care for themselves and function within a community. With appropriate training, they can secure employment, often in the competitive job market, especially through supported work programs.

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What Should Be Included in Education Programs?

Since Down syndrome is identifiable at birth in the majority of cases, an early intervention program, either in the home or in a center-based setting, affords both the child and the parent an opportunity to learn and grow. The Education of All Handicapped Children Act (P.L. 94–142) and the infant/toddler focus of P.L. 99–457, ensure each child the right to a free appropriate public education program until age 21.

The content of the school program should be closely aligned to the child's immediate needs and prior experiences. The curriculum should focus on communication skills, social skills, self-help skills, motor development, coping successfully with the physical environment, the enrichment of sensory experiences, functional academics, and daily living skills. It should be organized around long-term skill sequences designed to facilitate the acquisition of necessary independent functioning skills. As an adult, the individual should have the skills necessary to live in the community and secure vocational training and/or employment.

Rates of development vary from individual to individual; however, as with any person, education, stimulation, and the opportunity to participate in a variety of experiences facilitate all areas of development and assist the individual in becoming more able to deal with the daily environment in a meaningful way.

Can Down Syndrome Be Diagnosed Before Birth?

Down syndrome is the most common of the forms of mental retardation that can be identified during the prenatal and perinatal periods. Amniocentesis, which involves inserting a needle through the mother's abdomen and withdrawing amniotic fluid (fluid surrounding the fetus), can be performed around the 14th week of pregnancy. The fluid can be subjected to chemical tests and the chromosomes analyzed. In this manner, Down syndrome can be detected. Amniocentesis is often recommended if there is a history of Down syndrome in the family or if the woman is over 35 years of age. Alternatives to amniocentesis such as chorionic villus sampling are currently being investigated.

Resources

Association for Retarded Citizens—United States, P.O. Box 6109, Arlington, TX 76005 (1-800-433-5255).

National Down Syndrome Congress, 1800 Dempster Street, Park Ridge, IL 60068-1146 (1-800-232-6372) (312-823-7550).

National Down Syndrome Society, 141 5th Ave., New York, NY 10010 (212-460-9330).

The National Information Center for Handicapped Children and Youth, Park Place Bldg., Suite 1100, 7926 Jones Branch Drive, McLean, VA 22101 (703-893-6061).

Down Syndrome: A Resource Handbook (1988), by Carol Tingey, Editor. College Hill/Little Brown, 4284 41st Street, San Diego, CA 92105.

An Overview of Down Syndrome (1986) by Siegfried M. Pueschel. Available for $3.00 from ARC—United States, P.O. Box 6109, Arlington, TX 76005.

The Young Person with Down Syndrome: Transition From Adolescence to Adulthood (1988), by Siegfried M. Pueschel, Editor. Paul H. Brookes Publishing Company, P.O. Box 10624, Baltimore, MD 21285.

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